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CASE REPORT - PAEDIATRIC NEUROLOGY

Vitamin responsive bulbar palsy in an infant.

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Abstract

A seven months old infant presented with swallowing difficulty, poor cry and pooling of secretions of the throat as progressive bulbar palsy. She responded to riboflavin treatment and continued to improve.

Key-words: bulbar palsy, riboflavin, hypotonia, respiratory distress
Key Messages: riboflavin responsive bulbar palsy.

INTRODUCTION:

Vitamins used in clinical practice as food supplements sometimes act as cofactors for catalysis of many metabolic process in the body. A few neurological disorders, including seizures have been proved to be treated with vitamins in children. Progressive bulbar palsy in an infant is a severe debilitating illness with high morbidity. Many of the aetiologies of the disorder seems to be mostly neurodegenerative and have poor outcome. Fazio Londae disease, a similar rare disorder with progressive bulbar paralysis and respiratory failure, is found to be due to mutation of SLC52A3 gene which encodes the intestinal (hRFT2) riboflavin transporter in few children. They respond very well to prompt riboflavin therapy as this child we described and thus emphasizing this entity of riboflavin responsive neurological disorder as a unique vitamin responsive problem.

CASE HISTORY:

A 7 months old female infant presented with respiratory distress, difficulty in swallowing milk with poor cry. She was born as the first child to non consanguineous young parents as term SVD with no adverse perinatal events except transient neonatal jaundice and was discharged home on the 3rd day of life. Her developmental mile stones were normal except she had weak cry and poor neck muscle control. Mother noticed that ever since she had an upper respiratory catarrh, 3 weeks ago, she had been unwell with poor feeding, weak cough and breathlessness. She sought medical advice and the baby was treated for broncho pneumonia. Even after discharge from hospital after a few days, she continued to be unwell with feeding difficulty, pooling of secretions in the throat and husky cry. In the following week, she developed nasal regurgitation and increasing respiratory distress with nasal twang of the voice and was admitted to the intensive care unit as bulbar paralysis; underwent emergency endo tracheal intubation and connected to mechanical ventilator. She had all the primary vaccinations till date and there was no family history of any neurological illness. On examination, she was afebrile, stable vital signs, normal eye movements, bilateral facial paresis with absence of naso-labial folds and palatal palsy with poor gag reflex and palatal movements with pooling of saliva in the pharynx. She also had inconsistent tongue fasciculation, marked head lag, generalized hypotonia, norma power in all limbs. retained deep tendon reflexes and normal sensation all over the body. She could fix and follow light with normal optic fundi and intact hearing. Basic blood and biochemical workup including throat swab for diphtheria were done and reported normal.Baseline metabolic screening namely, ammonia, lactate, blood gases were normal including creatinine kinase (CK), thyroid profile, blood sugar, calcium and magnesium. X ray chest revealed bilateral pneumonic infiltration and was treated with antibiotics, chest physiotherapy and intravenous fluids. Several attempts to extubate the child were futile and so she underwent extensive

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investigations. MR brain and cervical spine were normal and metabolic screening with tandom mass spectroscopy, vitamin B 12 levels, survival motor neuron gene PCR (SMN gene) for spinal muscular atrophy were all negative. Detailed nerve conduction study revealed normal conduction velocities and F waves, normal sensory potentials; needle electromyography (EMG) showed fibrillation potentials over deltoid, biceps and triceps on both sides revealing axonal neuropathy (anterior horn cell). Parents refused for muscle biopsy and csf study to this child. With the limited available clinical picture and electro-physiological investigations she was diagnosed as neurogenic bulbospinal amyotrophy probably due to anterior horn cell disease. Following possibilities were put forth: spinal muscular atrophy (SMN gene mutation negative), hereditary motor sensory neuropathy type 1(nerve conduction study normal). Brown- Vialetto -Van Laere syndrome (normal hearing), Tay Sach's disease (absent startle myoclonus, cherry red spot) Fazio Londe disease.With the above differential diagnosis, she child could be suffering from progressive bulbar palsy of Fazio Londe disease ; a trial of riboflavin treatment at a dose of 15 mg per Kg body weight was given. Surprisingly, she showed dramatic improvement in 2 weeks time and successful extubation was done on the 25th day. Riboflavin was then continued on 10 mg per Kg weight of the child and discharged from hospital with the same medication. She was reviewed after 3 months; she started to swallow liquids as well semi solid food with no nasal regurgitation, could hold the neck and could sit and stand with support. She is still on the above treatment and advised regular follow up.

DISCUSSION:

Fazio Londe disease is a rare neurological disorder presenting with progressive bulbar palsy with respiratory failure. (1) It was once considered to have an unrelenting course, is now found to be due to mutations in the SLC52A3 gene which encodes the intestinal (hRFT2) riboflavin transporter in some children (2). Brown-Vialetto-Van Laere syndrome is similar rare neurological disorder with sensory-neural deafness, presenting at all ages with bulbar palsy and respiratory compromise and Fazio-Londe syndrome is considered to be the same disease entity presenting in children with normal hearing. (3)Appropriate diagnosis requires mutation analysis of allthe related three transporter genes of the above. (4) Because of the striking and often lifesaving effects of riboflavin supplementation it is highly advisable to start treatment immediately without awaiting results of mutation analysis. Thus despite lack of availability of geneticanalysis, a trail riboflavin therapy, administered to our child, on the basis of clinical symptoms resulted in a life saving effect.

CONCLUSION:

This case illustrates the rarity of presentation of progressive bulbar palsy of Fazio Lande disease in thischild on clinical and limited electrophysiological investigationand the beneficial life saving role of riboflavin treatment for this disorder. The point of philosophy is to start withsomething so simple ,and to end with something so surpriing that no one will believe it. Points to ponder!

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